☐1: AAA79207. inositol polyphos...[gi:1019103] BLink, Links

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LOCUS       AAA79207                 942 aa            linear   PRI 12-OCT-1995
DEFINITION  inositol polyphosphate 5-phosphatase.
ACCESSION   AAA79207
VERSION     AAA79207.1  GI:1019103
DBSOURCE    locus HUMINP5P accession M74161.1
KEYWORDS    .
SOURCE      Homo sapiens (human)
  ORGANISM  Homo sapiens
            Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
            Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.
REFERENCE   1  (residues 1 to 942)
  AUTHORS   Jefferson,A.B. and Majerus,P.W.
  TITLE     Properties of type II inositol polyphosphate 5-phosphatase
  JOURNAL   J. Biol. Chem. 270 (16), 9370-9377 (1995)
  MEDLINE   95238452
   PUBMED   7721860
COMMENT     Method: conceptual translation.
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      841 slpepvicys tyhnclecsg nytaskqvis tlpifhknvf hylmaflrel lknsaknhld
      901 enilasifgs lllrnpaghq kldmtekkka qefihqflcn pl
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Revised: July 5, 2002.

1: Hum Mol Genet 1995 Dec;4(12):2245-50 Related Articles, Links

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Related Resources

Lowe syndrome, a deficiency of phosphatidylinositol 4,5-bisphosphate 5-phosphatase in the Golgi apparatus.

Suchy SF, Olivos-Glander IM, Nussabaum RL.

Laboratory of Genetic Disease Research, National Center for Human Genome Research, National Insititues of Health, Bethesda, Maryland 20892, USA.

The oculocerebrorenal syndrome of Lowe (OCRL) is an X-linked disorder characterized by congenital cataracts, renal tubular dysfunction and neurological deficits. The gene responsible for this disorder, OCRL-1, has been cloned and mutations identified in patients. The gene product (ocrl-1) has extensive sequence homology to a 75 kDa inositol polyphosphate 5-phosphatase. We report here that OCRL patients' fibroblasts show no abnormality in inositol polyphosphate 5-phosphatase activity, but are deficient in a phosphatidylinositol 4,5-bisphosphate [PtdIns(4,5)P2] 5-phosphatase activity localized to the Golgi apparatus. Direct biochemical diagnosis of this human disease should now be possible. PtdIns(4,5)P2 has been implicated in Golgi vesicular transport through its role in the regulation of ADP-ribosylation factor, phospholipase D and actin assembly in the cytoskeleton. The regulation of PtdIns(4,5)P2 levels by PtdIns(4,5)P2 5-phosphatase may, therefore, be important in the modulation of Golgi vesicular transport. Given that the primary defect in OCRL is a deficiency of a Golgi PtdIns(4,5)P2 phosphatase, we hypothesize that the disorder results from dysregulation of Golgi function and in this way causes developmental defects in the lens and abnormal renal and neurological function.

PMID: 8634694 [PubMed - indexed for MEDLINE]

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☐1: AAB03216. phosphatidylinosi...[gi:1399105] BLink, Links

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LOCUS       AAB03216                 397 aa            linear   PRI 29-JUN-1996
DEFINITION  phosphatidylinositol (4,5)bisphosphate 5-phosphatase homolog.
ACCESSION   AAB03216
VERSION     AAB03216.1  GI:1399105
DBSOURCE    locus HSU45975 accession U45975.1
KEYWORDS    .
SOURCE      Homo sapiens (human)
  ORGANISM  Homo sapiens
            Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
            Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.
REFERENCE   1  (residues 1 to 397)
  AUTHORS   Nussbaum,R.L.
  TITLE     Direct Submission
  JOURNAL   Submitted (11-JAN-1996) Robert L. Nussbaum, NCHGR, NIH, 49 Convent
            Drive, Bethesda, MD 20892, USA
COMMENT     Method: conceptual translation.
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                     and R15390"
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     CDS             1..397
                     /coded_by="U45975.1:<1..1194"
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       61 sakkrkpawt drilwkvkap gggpspsgrk shrlqvtqhs yrshmeytvs dhkpvaaqfl
      121 lqfafrddmp lvrlevadew vrpeqavvry rmetvfarss wdwiglyrvg frhckdyvay
      181 vwakhedvdg ntyqvtfsee slpkghgdfi lgyyshnhsi ligitepfqi slpsselass
      241 stdssgtsse geddstlell apksrspspg kskrhrsrsp glarfpglal rpssrerrga
      301 srspspqsrr lsrvapdrss ngssrgssee gpsglpgpwa fppavprslg llpalrletv
      361 dpggggswgp drealapnsl spspqghrgl eegglgp
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Revised: July 5, 2002.

Sequence Revision History

Entrez Protein

Sequence Revision History				
Query	**GI**	**Version**	**Update Date**	**Status**
1420920	13254464	2	Mar 8 2001 17:16	Live
1420920	13254464	2	Mar 8 2001 17:15	Dead
1420920	1420920	1	Jul 16 1996 0:12	Dead

SeqId gi|1420920 was first seen at NCBI on Jul 16 1996 0:12

Related resources

Revised: July 5, 2002.

☐ 1: AAB03839

gb|AAB03839.1|[1420920]
The entry was deleted. See revision history for details.

Related resources

Revised: July 5, 2002.